

# Humangenetik Human Genetics Génétique humaine

---

## Editorial Board

**P. E. Becker**, Göttingen  
**A. G. Motulsky**, Seattle  
**J. W. Snyder**, Heidelberg  
**E. Vogel**, Heidelberg  
**G. G. Wendt**, Marburg

## Advisory Board

**S. Anders**, Groningen  
**J. Baitsch**, Ulm  
**A. G. Bearn**, New York  
**J. Bickel**, Heidelberg  
**J. P. Bochkov**, Moskau  
**D. Bootsma**, Rotterdam  
**H. Degenhardt**, Frankfurt/M.  
**V. Fuhrmann**, Giessen  
**J. Grüneberg**, London  
**J. Hassenstein**, Freiburg i. Br.

**K. Hirschhorn**, New York  
**W. Jaeger**, Heidelberg  
**D. Klein**, Genève  
**E. Krah**, Heidelberg  
**W. Krone**, Ulm  
**H. Lehmann**, Cambridge  
**W. Lenz**, Münster/W.  
**V. A. McKusick**, Baltimore  
**M. Mikkelsen**, Glostrup  
**H. Nachtsheim**, Berlin

**K. Patau**, Madison  
**A. Prader**, Zürich  
**H. Ritter**, Tübingen  
**C. Ropartz**, Bois-Guillaume  
**W. Schmid**, Zürich  
**W. J. Schull**, Ann Arbor  
**H. G. Schwarzscher**, Wien  
**C. Stern**, Berkeley  
**H. E. Sutton**, Austin  
**U. Wolf**, Freiburg i. Br.

Band 27 · 1975



Springer-Verlag · Berlin · Heidelberg · New York

The exclusive copyright for all languages and countries, including the right for photomechanical and any other reproduction, also in microform, is transferred to the publisher.

The use in this journal of registered or trade names, trademarks etc. without special acknowledgement does not imply that such names, as defined by the relevant protection laws, may be regarded as unprotected and thus free for general use.

Alle Rechte, einschließlich das der Übersetzung in fremde Sprachen und das der fotomechanischen Wiedergabe oder einer sonstigen Vervielfältigung, auch in Mikroform, vorbehalten. Jedoch wird gewerblichen Unternehmen für den innerbetrieblichen Gebrauch nach Maßgabe des zwischen dem Börsenverein des Deutschen Buchhandels e.V. und dem Bundesverband der Deutschen Industrie abgeschlossenen Rahmenabkommens die Anfertigung einer fotomechanischen Vervielfältigung gestattet. Wenn für diese Zeitschrift kein Pauschalabkommen mit dem Verlag vereinbart worden ist, ist eine Wertmarke im Betrage von DM 0,40 pro Seite zu verwenden.

*Der Verlag läßt diese Beträge den Autorenverbänden zufließen.*

Die Wiedergabe von Gebrauchsnamen, Handelsnamen, Warenbezeichnungen usw. in dieser Zeitschrift berechtigt auch ohne besondere Kennzeichnung nicht zu der Annahme, daß solche Namen im Sinne der Warenzeichen- und Markenschutz-Gesetzgebung als frei zu betrachten wären und daher von jedermann benutzt werden dürften.

Springer-Verlag Berlin · Heidelberg · New York

Printed in Germany by J. P. Peter, Gebr. Holstein, Rothenburg o. d. Thür.

© by Springer-Verlag Berlin · Heidelberg 1975

# Contents

## Reviews

Zeuthen, J.: Heterokaryons in the Analysis of Genes and Gene Regulation . . . . .	275
---	-----

## Original Investigations · Short Communications

Abele, R., s. Schneider, P., <i>et al.</i> . . . . .	217
Agarwal, D. P., s. Willers, I., <i>et al.</i> . . . . .	323
Albert, E. D., s. Rittner, C., <i>et al.</i> . . . . .	173
Ananthakrishnan, R., s. Schneider, P., <i>et al.</i> . . . . .	217
Arias, S., Mota, M., de Yáñez, A., Bolívar, M.: Probable Loose Linkage between the ABO Locus and Waardenburg Syndrome Type I . . . . .	145
Arndt-Hanser, A., s. Walter, H., <i>et al.</i> . . . . .	129
Atkin, J., s. Rundle, A. T., <i>et al.</i> . . . . .	15
Becker, J., s. Mortier, W., <i>et al.</i> . . . . .	199
Benkmann, H.-G., Goedde, H. W.: Esterase D-Polymorphism in Assam by Cellulose Ace- tate Electrophoresis . . . . .	343
Bentegeat, J., s. Serville, F., <i>et al.</i> . . . . .	49
Berg, K., s. Kamel, R. A., <i>et al.</i> . . . . .	53
Bergstrom, T., s. Magenis, R. E., <i>et al.</i> . . . . .	91
Bethge, R., s. Bissbort, S., <i>et al.</i> . . . . .	57
Bissbort, S., Kömpf, J., Bethge, R., Gussmann, S.: Population Genetics of Human Red Cell Phosphoglucose mutase Isozyme PGM <sub>3</sub> (E.C.:2.7.5.1). Gene Frequencies in Southwestern Germany . . . . .	57
Bissbort, S., s. Kömpf, J., <i>et al.</i> . . . . .	141
Boivin, P., s. Kahn, A., <i>et al.</i> . . . . .	247
Bolívar, M., s. Arias, S., <i>et al.</i> . . . . .	145
Broustet, A., Serville, F., Roger, P., Gachet, M.: X Monosomy and 21 Trisomy in a Sibship . . . . .	333
Bühler, E. M., s. Frey, R. O., <i>et al.</i> . . . . .	81
Bühler, U. K., s. Frey, R. O., <i>et al.</i> . . . . .	81
Chrz, R., s. Michalová, K., <i>et al.</i> . . . . .	157
Coldwell, J. G., s. Say, B. . . . .	231
Crossen, P. E.: Giemsa Banding Patterns in Chronic Lymphocytic Leukaemia . . . . .	151
Dieker, P., s. Utermann, G., <i>et al.</i> . . . . .	185
Dossetor, J. B., s. Segal, D. J., <i>et al.</i> . . . . .	45
Dutrillaux, B., Gueguen, J.: Etude méiotique et mitotique dans un cas de translocation t(5;Y) . . . . .	241
Enzenauer, J., s. Matz, D., <i>et al.</i> . . . . .	309
Frey, R. O., Bühler, E. M., Bühler, U. K., Stalder, G. R.: 45,X/46,XYq dic-Geschlechts- chromosomenmosaik . . . . .	81
Gachet, M., s. Broustet, A., <i>et al.</i> . . . . .	333
Gaensslen, R. E., s. Welch, S. G., <i>et al.</i> . . . . .	59
Gerhard, L., s. Mortier, W., <i>et al.</i> . . . . .	199
Giblett, E. R., s. Stamatoyannopoulos, G., <i>et al.</i> . . . . .	23
Goedde, H. W., s. Benkmann, H.-G. . . . .	343
Goedde, H. W., s. Willers, I., <i>et al.</i> . . . . .	323
Grimm, T., s. Langenbeck, U., <i>et al.</i> . . . . .	315
Gropp, A., s. Kucherlapati, R. S., <i>et al.</i> . . . . .	9
Grosse-Wilde, H., s. Rittner, C., <i>et al.</i> . . . . .	173
Gueguen, J., s. Dutrillaux, B., <i>et al.</i> . . . . .	241
Gumbel, B., s. Walter, H., <i>et al.</i> . . . . .	129
Gupta, S. C., s. Mehrotra, T. N., <i>et al.</i> . . . . .	347



Gussmann, S., s. Bissbort, S., <i>et al.</i> . . . . .	57
Gussmann, S., s. Kömpf, J., <i>et al.</i> . . . . .	141
Hansson, A., s. Mikkelsen, M., <i>et al.</i> . . . . .	303
Hecht, F., s. Magenis, R. E., <i>et al.</i> . . . . .	91
Hepp, R., Krüger, J., Kurzen, S., Rupp, H., Vogel, F.: ABO Blood Groups and Chicken Pox . . . . .	329
Hilwig, I., s. Kucherlapati, R. S., <i>et al.</i> . . . . .	9
Hobolth, N., s. Mikkelsen, M., <i>et al.</i> . . . . .	303
Jacobsen, P., s. Mikkelsen, M., <i>et al.</i> . . . . .	303
Kahn, A., Norht, L., Messer, J., Boivin, P.: G-6PD "Ankara". A New G-6PD Variant with Deficiency Found in a Turkish Family . . . . .	247
Kamel, R. A., Berg, K., Schwarzfischer, F., Wischerat, H.: First Determination of the Isozyme Patterns of Phosphoglycerate Mutases (E.C.2.7.5.3) and Phosphoglycerate Kinases (E.C.2.7.2.3) in Human Tissues . . . . .	53
Kömpf, J., s. Bissbort, S., <i>et al.</i> . . . . .	57
Kömpf, J., Bissbort, S., Gussmann, S., Ritter, H.: Polymorphism of Red Cell Glyoxalase I (E.C.: 4.4.1.5). A New Genetic Marker in Man. Investigation of 169 Mother-Child Combinations . . . . .	141
Krausz, T., s. Sellyei, M., <i>et al.</i> . . . . .	339
Krüger, J., s. Hepp, R., <i>et al.</i> . . . . .	329
Kucherlapati, R. S., Hilwig, I., Gropp, A., Ruddle, F. H.: Mammalian Chromosome Identification in Interspecific Hybrid Cells using "Hoechst 33258" . . . . .	9
Kurzen, S., s. Hepp, R., <i>et al.</i> . . . . .	329
Langenbeck, U., Grimm, T., Rüdiger, H. W., Passarge, E.: Heterozygote Tests and Genetic Counseling in Maple Syrup Urine Disease. An Application of Bayes' Theorem . . . . .	315
Langer, K. H., s. Utermann, G., <i>et al.</i> . . . . .	185
Lehmann, H., s. Mehrotra, T. N., <i>et al.</i> . . . . .	347
Lorenz, H., s. Rittner, C., <i>et al.</i> . . . . .	173
Lovrien, E., s. Magenis, R. E., <i>et al.</i> . . . . .	91
Magenis, R. E., Overton, K., Wyandt, H., Bergstrom, T., Hecht, F., Lovrien, E.: Exclusion Gene Mapping Utilizing Patients with Chromosome Imbalance: The HL-A System as a Prototype . . . . .	91
Málková, J., s. Michalová, K., <i>et al.</i> . . . . .	157
Manso, C., s. Martins Pereira, T. . . . .	137
Martins Pereira, T., Manso, C.: Immunoglobulin Allotypes in Portugal . . . . .	137
Mattevi, M. S., Salzano, F. M.: Senescence and Human Chromosome Changes . . . . .	1
Matz, D., Enzenauer, J., Menne, F.: Über einen Fall von atypischer Galaktosämie . . . . .	309
McCoy, E. E., s. Segal, D. J., <i>et al.</i> . . . . .	45
Mehrotra, T. N., Gupta, S. C., Sinha, R., Lehmann, H., Wiltshire, B. G.: Haemoglobin Norfolk in Nepali Gorkhas . . . . .	347
Menne, F., s. Matz, D., <i>et al.</i> . . . . .	309
Menzel, H. J., s. Utermann, G., <i>et al.</i> . . . . .	185
Messer, J., s. Kahn, A., <i>et al.</i> . . . . .	247
Michaelis, E., s. Mortier, W., <i>et al.</i> . . . . .	199
Michalová, K., Málková, J., Chrz, R.: Two Cases of C-Group Balanced Translocations . . . . .	157
Mikkelsen, M., Hansson, A., Jacobsen, P., Hobolth, N.: Translocation (13q21q). Four Generation Family Study with Analysis of Satellite Associations, Fluorescent Markers, and Prenatal Diagnosis . . . . .	303
Mills, P. R., s. Welch, S. G., <i>et al.</i> . . . . .	59
Mortier, W., Michaelis, E., Becker, J., Gerhard, L.: Centronucleäre Myopathie mit autosomal dominantem Erbgang . . . . .	199
Mota, M., s. Arias, S., <i>et al.</i> . . . . .	145
Netzel, B., s. Rittner, C., <i>et al.</i> . . . . .	173
Norht, M. L., s. Kahn, A., <i>et al.</i> . . . . .	247
Obe, G., s. Sperling, K., <i>et al.</i> . . . . .	227
Overton, K., s. Magenis, R. E., <i>et al.</i> . . . . .	91
Pabst, H. F., s. Segal, D. J., <i>et al.</i> . . . . .	45

Passarge, E., s. Langenbeck, U., <i>et al.</i> . . . . .	315
Pihar, O.: Red Blood Cell Acid Phosphatase: Ambiguity in Phenotype and Activity Estimations in the Proof of the "Single Allele" States . . . . .	235
Raffa, M.-A., s. Walter, H., <i>et al.</i> . . . . .	129
Riehm, H., s. Sperling, K., <i>et al.</i> . . . . .	227
Ritter, H., s. Kömpf, J., <i>et al.</i> . . . . .	141
Rittner, C., Grosse-Wilde, H., Rittner, B., Netzel, B., Scholz, S., Lorenz, H., Albert, E. D.: Linkage Group HL-A-MLC-BF (Properdin Factor B). The Site of the Bf Locus at the Immunogenetic Linkage Group on Chromosome 6 . . . . .	173
Rittner, B., s. Rittner, C., <i>et al.</i> . . . . .	173
Roger, P., s. Broustet, A., <i>et al.</i> . . . . .	333
Ruddle, F. H., s. Kucherlapati, R. S., <i>et al.</i> . . . . .	9
Rüdiger, H. W., s. Langenbeck, U., <i>et al.</i> . . . . .	315
Rundle, A. T., Atkin, J., Sudell, B.: Serum and Tissue Proteins in Tuberos Sclerosis. I. Serum and Red-Cell Polymorphic Systems . . . . .	15
Rupp, H., s. Hepp, R., <i>et al.</i> . . . . .	329
Salzano, F. M., s. Mattevi, M. S. . . . . .	1
Sanchez, O., s. Yunis, J. J. . . . . .	167
Say, B., Coldwell, J. G.: Hereditary Defect of the Sacrum . . . . .	231
Schlaut, J. W., s. Segal, D. J., <i>et al.</i> . . . . .	45
Schloot, W., s. Willers, I., <i>et al.</i> . . . . .	323
Schneider, P., Ananthakrishnan, R., Walter, H., Xirotiris, N., Abele, R.: Enzyme Poly- morphisms and Haemoglobin Variants in Greeks . . . . .	217
Scholz, S., s. Rittner, C., <i>et al.</i> . . . . .	173
Schwarzfischer, F., s. Kamel, R. A., <i>et al.</i> . . . . .	53
Segal, D. J., Schlaut, J. W., Pabst, H. F., McCoy, E. E., Dossetor, J. B.: HL-A Fre- quencies in Down's Syndrome . . . . .	45
Seidel, H., s. Zankl, H., <i>et al.</i> . . . . .	119
Sellyei, M., Vass, L., Krausz, T.: Non-Random Appearance of Y-Chromatin-Like Fluorescence in the Nuclei of Thyroid and Brain and Its Chromosomal Background . . . . .	339
Serville, F., Bentegeat, J., Verger, P.: Elliptocytosis: Linkage Study in a Family . . . . .	49
Serville, F., s. Broustet, A., <i>et al.</i> . . . . .	333
Shiraishi, Y.: Cytogenetic Studies in 12 Patients with Itai-Itai Disease . . . . .	31
Singh, S., s. Willers, I., <i>et al.</i> . . . . .	323
Sinha, R., s. Mehrotra, T. N., <i>et al.</i> . . . . .	347
Spaeter, M.: Nichtzufällige Verteilung homologer Chromosomen (Nr. 9 und YY) in Interphasekernen menschlicher Fibroblasten . . . . .	111
Sperling, K., Wegner, R.-D., Riehm, H., Obe, G.: Frequency and Distribution of Sister- Chromatid Exchanges in a Case of Fanconi's Anemia . . . . .	227
Stalder, G. R., s. Frey, R. O., <i>et al.</i> . . . . .	81
Stamatoyannopoulos, G., Thomakos, A., Giblett, E. R.: Red Cell Enzyme Poly- morphisms in the Greek Populations . . . . .	23
Sudell, B., s. Rundle, A. T., <i>et al.</i> . . . . .	15
Thomakos, A., s. Stamatoyannopoulos, G., <i>et al.</i> . . . . .	23
Utermann, G., Menzel, H. J., Langer, K. H., Dieker, P.: Lipoproteins in Lecithin- Cholesterol-Acyltransferase (LCAT)-Deficiency. II. Further Studies on the Abnormal High-Density-Lipoproteins . . . . .	185
Vass, L., s. Sellyei, M., <i>et al.</i> . . . . .	339
Verger, P., s. Serville, F., <i>et al.</i> . . . . .	49
Vogel, F., s. Hepp, R., <i>et al.</i> . . . . .	329
Wahlström, J.: Three Cases of Minor Chromosomal Aberrations Discovered by Prenatal Chromosome Determination . . . . .	223
Walter, H., Arndt-Hanser, A., Raffa, M.-A., Gumbel, B.: On the Distribution of Some Genetic Markers in Libya . . . . .	129
Walter, H., s. Schneider, P., <i>et al.</i> . . . . .	217
Wegner, R.-D., s. Sperling, K., <i>et al.</i> . . . . .	227



Welch, S.: Population and Family Studies on Carbonic Anhydrase II Polymorphism in Gambia, West Africa . . . . .	163
Welch, S. G., Mills, P. R., Gaensslen, R. E.: Phenotypic Distributions of Red Cell Glutamate-Pyruvate Transaminase (E.C. 2.6.1.2) Isoenzymes in British and New York Populations . . . . .	59
Willers, I., Agarwal, D. P., Singh, S., Schloot, W., Goedde, H. W.: Rapid Determination of Hypoxanthine-Guanine-Phosphoribosyl Transferase in Human Fibroblasts and Amniotic Cells . . . . .	323
Wiltshire, B. G., s. Mehrotra, T. N., <i>et al.</i> . . . . .	347
Wischerat, H., s. Kamel, R. A., <i>et al.</i> . . . . .	53
Wyandt, H., s. Magenis, R. E., <i>et al.</i> . . . . .	91
Xirotiris, N., s. Schneider, P., <i>et al.</i> . . . . .	217
Yáñez, de, A., s. Arias, S., <i>et al.</i> . . . . .	145
Yunis, J. J., Sanchez, O.: The G-Banded Prophase Chromosomes of Man . . . . .	167
Zang, K. D., s. Zankl, H., <i>et al.</i> . . . . .	119
Zankl, H., Seidel, H., Zang, K. D.: Cytological and Cytogenetical Studies on Brain Tumors. V. Preferential Loss of Sex Chromosomes in Human Meningiomas . . . . .	119

#### *Clinical Case Reports*

Balíček, P., Žižka, J., Lichý, J.: A Case of Trisomy 9p in a Family with Translocation 9/15 . . . . .	353
David, T. J., Jones, A. J.: Trisomy 21 and Trisomy 18 in Half-Siblings . . . . .	351
Fujimoto, A., Wilson, M. G., Towner, J. W.: Familial Inversion of Chromosome No. 8. An Affected Child and a Carrier Fetus . . . . .	67
Kužerová, M., Polívková, Z., Pokorná, M.: Deletion of Long Arms of Chromosome 13 . . . . .	255
Málková, J., Michalová, K., Chrz, R., Kobilková, J., Motlík, K., Stárka, L.: Dicentric Yp Chromosome in a Patient with the Gonadal Dysgenesis and Gonadoblastoma . . . . .	251
Schmid, W., Mühlethaler, J. P., Briner, J., Knechtli, H.: Ring Chromosome in a Polymalformed Anencephalic . . . . .	63
Stoll, C., Juif, J. G., Luckel, J. C., Lausecker, C.: Ring Chromosome 15:r (15). Identification by R Banding . . . . .	259
Stoll, C., Levy, J.-M., Champy, M.: Balanced Familial Translocation t(5;19) (q12;p or q11) with Phenotypical Abnormalities in a Girl . . . . .	263
Stoll, C., Levy, J.-M., Gardea, A.: Trisomy 9p in a Girl whose Mother has a Translocation t(9;20) (q12;p13) . . . . .	269

#### *Letters to the Editors*

Kühnl, P., Nowicki, L., Spielmann, W.: Schlußwort . . . . .	77
Ritter, H.: Diskussionsbemerkung zur Arbeit: Untersuchungen zum Polymorphismus der Galaktose-1-Phosphat-Uridyl-Transferase (EC: 2.7.7.12) mittels Agarosegelelektrophorese von P. Kühnl, L. Nowicki und W. Spielmann. Humangenetik 24, 227—230 (1974) . . . . .	75

## Notes on Preparation of Illustrations

---

*Selection of illustration material:* In order to obtain the best results in reproduction, to avoid delays during production and hence unnecessary costs, we ask authors to note the following points when selecting and preparing illustration copy.

### 1. Half-tones (photographs, photomicrographs, X-rays, instrument traces etc.)

- Send only good, well-contrasted glossy prints of the original negative; prints should be trimmed at right angles; send contact copies of X-rays — if these are not available, the actual X-ray films.
- Mark or trim off marginal portions which are not required (at right angles, please).
- State scale of reduction, if any, with due allowance for the format of the printed page (print area).
- Group figures into whole-page plates; see that they match in the proposed scale of reduction.
- With X-rays, in particular, mark the significant portions on the back of the copy, or on a cover sheet.
- Enter inscriptions, marker lines etc. neatly and in the appropriate size, either on the photograph itself or on a cover sheet.

### 2. Line drawings

- State final size of illustration, with due allowance for print area.
- The ideal is for drawings to be twice the final size and executed in indelible black ink.

Important points to note: thickness of lines, size of inscriptions, size of measuring points, adequate spacing of shaded and dotted areas.

Words should be in upper and lower case characters (not block capitals).

Example showing the effect of reduction  $\times \frac{1}{2}$ .

ABCDEFGHIJKLMNOPQRSTUVWXYZ  
abcdefghijklmnopqrstuvwxyzß  
1234567890  
(!:"'+=,;x?%)

ABCDEFGHIJKLMNOPQRSTUVWXYZ  
abcdefghijklmnopqrstuvwxyzß  
1234567890  
(!:"'+=,;x?%)

---

Some more examples see overleaf



